CLAIMS

What is claimed is:

1. A method for detection of a variant Cayman ataxia polypeptide or nucleic acid sequence in a subject, comprising:

a) providing a biological sample from a subject, wherein said biological sample comprises a Cayman ataxia polypeptide or nucleic acid; and

b) detecting the presence or absence of a variant Cayman ataxia polypeptide or nucleic acid in said biological sample.

2. The method of claim 1, wherein said variant Cayman ataxia polypeptide is a variant of SEQ ID NO:4.

3. The method of claim 2, wherein said variant Cayman ataxia polypeptide comprises SEQ ID NO:9.

4. The method of claim 1, wherein said variant Cayman ataxia nucleic acid is a variant of a sequence selected from the group consisting of SEQ ID NOs:3 and 11.

5. The method of claim 4, wherein said variant Cayman ataxia nucleic acid is selected from the group consisting of SEQ ID NOs: 8 and 10.

6. The method of claim 1, wherein the presence of said variant Cayman ataxia polypeptide or nucleic acid is indicative of Caymans ataxia in said subject.

7. The method of claim 1, wherein the presence of said variant Cayman ataxia polypeptide or nucleic acid is indicative of said subject being a Cayman ataxia carrier.

8. The method of claim 1, wherein the presence of said variant Cayman ataxia polypeptide or nucleic acid is indicative of a disorder selected from the group consisting of ataxia, myoclonus, dystonia, epilepsy, and nystagmus in said subject.

9. The method of claim 1, wherein said biological sample is selected from the group consisting of a blood sample, a tissue sample, a urine sample, a saliva sample, and an amniotic fluid sample.

10. The method of claim 1, wherein said subject is selected from the group consisting of an embryo, a fetus, a newborn animal, a young animal, and an adult animal.

11. The method of claim 10, wherein said animal is a human.

12. The method of claim 10, wherein said human is an adult female of child-bearing age.

13. The method of claim 1, wherein said detecting comprises differential antibody binding.

14. The method of claim 1, wherein said detection comprises a Western blot.

15. The method of claim 1, wherein said detection comprises a nucleic acid detection method selected from the group consisting of nucleic acid sequencing, polymerase chain reaction, hybridization, denaturing high pressure liquid chromatography, mass spectrometry, and enzymatic detection.

16. A kit comprising a reagent for detecting the presence or absence of a variant Cayman ataxia nucleic acid or polypeptide in a biological sample.

17. The kit of claim 16, further comprising instruction for using said kit for said detecting the presence or absence of a variant Cayman ataxia nucleic acid or polypeptide in a biological sample.

18. The kit of claim 16, wherein said instructions comprise instructions required by the U.S. Food and Drug Agency for *in vitro* diagnostic kits.

19. The kit of claim 16, further comprising instructions for diagnosing Caymans ataxia in said subject based on the presence or absence of said variant Cayman ataxia polypeptide.

20. The kit of claim 16, further comprising instructions for diagnosing Caymans ataxia carrier status in said subject based on the presence or absence of said variant Cayman ataxia polypeptide.

21. The kit of claim 16, further comprising instructions for diagnosing a disorder selected from the group consisting of ataxia, myoclonus, dystonia, epilepsy, and nystagmus in said subject based on the presence or absence of said variant Cayman ataxia polypeptide.

22. The kit of claim 16, wherein said reagent is one or more antibodies.

23. The kit of claim 16, wherein said reagents comprise reagents for performing a nucleic acid detection assay selected from the group consisting of nucleic acid sequencing, polymerase chain reaction, hybridization, denaturing high pressure liquid chromatography, mass spectrometry, and enzymatic detection.

24. The kit of claim 16, wherein said variant Cayman ataxia polypeptide is a variant of SEQ ID NO:4.

25. The kit of claim 24, wherein said variant Cayman ataxia polypeptide comprises SEQ ID NO:9.

26. The kit of claim 16, wherein said variant Cayman ataxia nucleic acid is a variant of a nucleic acid sequence selected from the group consisting of SEQ ID NOs: 3 and 11.

27. The kit of claim 26, wherein said variant Cayman ataxia nucleic acid is selected from the group consisting of SEQ ID NOs: 8 and 10.

28. The kit of claim 16, wherein said biological sample is selected from the group consisting of a blood sample, a tissue sample, a urine sample, a saliva sample, and an amniotic fluid sample.